

CLAIMS

WHAT IS CLAIMED IS:

- 1 1. A kit for determining whether a subject has, or is at risk of developing, colorectal cancer
2 wherein said kit is used to amplify and/or determine the molecular structure of at least a
3 portion of the MnSOD gene.
- 1 2. The kit of claim 1 further comprising a first and second oligonucleotide specific for SEQ
2 ID NO: 1.
- 1 3. The kit of claim 2 wherein said first and second oligonucleotides can be used to produce a
2 polynucleotide comprising a region of the MnSOD gene, said region including nucleotide
3 residue 351 of SEQ ID NO:1.
- 1 4. The kit of claim 2, wherein the oligonucleotides have a nucleotide sequence from about 15
2 to about 30 nucleotides.
- 1 5. The kit of claim 2, wherein the first and second oligonucleotides are labeled.
- 1 6. The kit of claim 2, wherein the first oligonucleotide is specific for the MnSOD Ala allele
2 and the second oligonucleotide is specific for the MnSOD Val allele.
- 1 7. The kit of claim 1 further comprising one or more oligonucleotide probes specific for the
2 MnSOD Ala allele and the MnSOD Val allele.
- 1 8. The kit of claim 7 wherein said probes are detectably labeled.
- 1 9. The kit of claim 8, wherein said probes are fluorescently labeled.
- 1 10. The kit of claim 9, wherein said probes are labeled with a quenching molecule.
- 1 11. The kit of claim 7, wherein said probes are bound to a surface.
- 1 12. The kit of claim 1 further comprising an allele specific endonuclease.

- 1 13. A method for determining whether a subject has, or is at a risk of developing, colorectal
2 cancer comprising determining the identity of the allelic variant of the MnSOD gene in a
3 nucleic acid obtained from the subject.

1 14. The method of claim 13 further comprising contacting the subject's sample nucleic acid
2 comprising the MnSOD gene with a probe or primer which hybridizes to the polymorphic
3 region of the mitochondrial targeting sequence of the MnSOD gene, said polymorphic
4 region including nucleotide 351 of SEQ ID NO:1.

1 15. The method of claim 13, wherein determining the identity of the allelic variant comprises
2 determining the identity of at least one nucleotide of the polymorphic region.

1 16. The method of claim 13, wherein determining the identity of the allelic variant comprises
2 performing a restriction enzyme site analysis.

1 17. The method of claim 13, wherein determining the identity of the allelic variant is carried out
2 by single-stranded conformation polymorphism.

1 18. The method of claim 13, wherein determining the identity of the allelic variant is carried out
2 by allele specific hybridization.

1 19. The method of claim 13, wherein determining the identity of the allelic variant is carried out
2 by primer specific extension.

1 20. The method of claim 13, wherein determining the identity of the allelic variant is carried out
2 by an oligonucleotide ligation assay.

1 21. The method of claim 13, wherein the MnSOD gene is a human MnSOD gene.

1 22. The method of claim 13, wherein the probe or primer has a nucleotide sequence from about
2 15 to about 30 nucleotides.

1 23. The method of claim 13, wherein the probe or primer is labeled.

1 24. A method for determining risk of colorectal cancer in a subject, comprising the steps of:
2 a. determining the base identity of a portion of genomic DNA from the subject's cell

3 sample, said genomic DNA comprising an MnSOD gene comprising a mitochondrial
targeting sequence, said portion corresponding to position 351 as defined in SEQ ID
5 NO:1 of said MnSOD gene in said mitochondrial targeting sequence; and
6 b. correlating said base identity with a risk for colorectal cancer.

- 1 25. The method of claim 24; wherein the base identity of position 351 is determined by
2 sequencing a portion of said mitochondrial targeting sequence of said MnSOD gene
3 containing said position 351.
- 1 26. The method of claim 24; wherein base identity of said position 351 is determined by
2 digesting said portion of the mitochondrial targeting sequence of said MnSOD gene with a
3 restriction endonuclease appropriate to determine the base identity of said position 351.
- 1 27. The method of claim 24; wherein said base identity is determined by examining an RNA
2 fraction from said subject's cell sample, whereby the identity of said genomic DNA at said
3 position 351 can be determined.
- 1 28. The method of claim 24; wherein a risk for developing colorectal cancer is assessed to be
2 greater than that of the unaffected relevant population when the base identity at said position
3 351 is homozygous for C.
- 1 29. The method of claim 28; wherein the age of the subject is less than about 35 years.
- 1 30. The method of claim 29; wherein the ethnicity of the subject is Hispanic.